AMENDMENTS

In the Specification

This application claims priority to United States Provisional Application Serial No. 60/331362 filed January 4, 2001 and sent bearing Express Mail Label EL 389 348 319 US to the United States Patent and Trademark Office on January 4, 2001.

Figure 1 depicts a gene and protein structure of *AIPL1*; ***a.*** *AIPL1* consists of six exons, with alternate polyadenylation sites in the 3' untranslated region, shown by arrows. Cys239Arg denotes the location of the TGC→CGC missense mutation in exon 5 of the RFS128 family. Trp278X denotes the location of the TGG→TGA nonsense mutation in exon 6 of the KC, MD, RFS127 and RFS121 families. Ala336Δ2 denotes the location of the 2 bp deletion in exon 6 of RFS121. Benign coding sequence substitutions identified were Phe37Phe (TTT/TTC; 0.98/0.02 frequency), Cys89Cys (TGC/TGT; 0.99/0.01), Asp90His (GAC/CAC; 0.84/0.16), Leu100Leu (CTG/CTA; 0.57/0.43) and Pro217Pro (CCG/CCA; 0.61/0.39) ***b.*** Protein sequence of AIPL1. The alignment demonstrates the high level of sequence conservation between rat AIPL1 (SEQ. ID NO. 81) and human AIPL1 (SEQ. ID NO. 72), and mouse AIP (SEQ. ID NO. 80) and human AIP (SEQ. ID NO. 79). Identical residues in the four sequences are noted with an asterisk; identical residues in three of the sequences are indicated with a period.;

Figure 9 depicts pedigrees of two families with probands heterozygous for the 12 bp *AIPL1* deletion, and representative electropherogram. The mutant allele was sucloned and sequenced to confirm size and sequence of deletion. Family members who have not been clinically examined, and, therefore, are of unknown phenotype are designated by an "?" within the symbol. (A) UTAD231, with the original diagnosis of "cone-rod dystrophy, possibly dominant". Two unaffected individuals in this family lack the 12 bp deletion (tgcagagccacc) (SEQ. ID NO. 82). (B) UTAD907, whose original diagnosis was "juvenile RP, possibly dominant"; and